



RNASEH2A gene

ribonuclease H2 subunit A

Normal Function

The *RNASEH2A* gene provides instructions for making one part (subunit) of a group of proteins called the RNase H2 complex. This complex is a ribonuclease, which means it is an enzyme that helps break down RNA, a chemical cousin of DNA. In particular, the RNase H2 complex helps break down molecules in which one strand of RNA is combined with one strand of DNA (RNA-DNA hybrids). These hybrids are formed during DNA copying (replication) and are found in all cells.

The RNase H2 complex is involved in DNA replication, error repair, and other cellular functions. Researchers believe that these additional functions may include helping to prevent inappropriate immune system activation.

Health Conditions Related to Genetic Changes

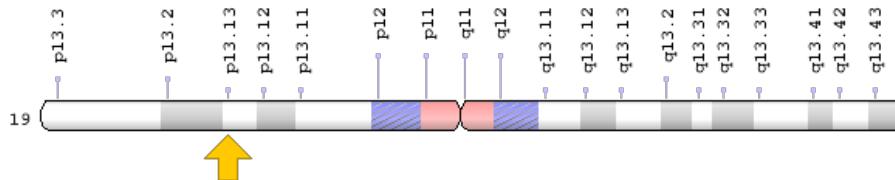
Aicardi-Goutieres syndrome

At least nine mutations in the *RNASEH2A* gene have been identified in people with Aicardi-Goutieres syndrome. These mutations likely result in a dysfunctional RNase H2 complex. The resulting disruption in cellular functions may lead to an accumulation of unneeded DNA and RNA in cells. These DNA and RNA molecules or fragments may be generated during the first stage of protein production (transcription), replication of cells' genetic material in preparation for cell division, DNA repair, cell death, and other processes. The unneeded DNA and RNA may be mistaken by cells for those of viral invaders, triggering immune system reactions that cause severe brain dysfunction (encephalopathy), skin lesions, and other signs and symptoms of Aicardi-Goutieres syndrome.

Chromosomal Location

Cytogenetic Location: 19p13.13, which is the short (p) arm of chromosome 19 at position 13.13

Molecular Location: base pairs 12,802,054 to 12,813,648 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AGS4
- aicardi-Goutieres syndrome 4 protein
- JUNB
- ribonuclease H2, large subunit
- ribonuclease H2, subunit A
- ribonuclease H1 large subunit
- ribonuclease H1 subunit A
- ribonuclease H1, large subunit
- RNase H(35)
- RNase H2 subunit A
- RNase H1 large subunit
- RNASEHI
- RNH2A_HUMAN
- RNHIA
- RNHL

Additional Information & Resources

GeneReviews

- Aicardi-Goutieres Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1475>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28RNASEH2A%5BTIAB%5D%29+OR+%28%28AGS4%5BTIAB%5D%29+OR+%28RNHL%5BTIAB%5D%29+OR+%28RNASEHI%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- RIBONUCLEASE H2, SUBUNIT A
<http://omim.org/entry/606034>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_RNASEH2A.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RNASEH2A%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18518
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10535>
- UniProt
<http://www.uniprot.org/uniprot/O75792>

Sources for This Summary

- Perrino FW, Harvey S, Shaban NM, Hollis T. RNaseH2 mutants that cause Aicardi-Goutieres syndrome are active nucleases. *J Mol Med (Berl)*. 2009 Jan;87(1):25-30. doi: 10.1007/s00109-008-0422-3. Epub 2008 Nov 26.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19034401>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2852111/>
- OMIM: RIBONUCLEASE H2, SUBUNIT A
<http://omim.org/entry/606034>

- Rice G, Patrick T, Parmar R, Taylor CF, Aeby A, Aicardi J, Artuch R, Montalto SA, Bacino CA, Barroso B, Baxter P, Benko WS, Bergmann C, Bertini E, Biancheri R, Blair EM, Blau N, Bonthon DT, Briggs T, Brueton LA, Brunner HG, Burke CJ, Carr IM, Carvalho DR, Chandler KE, Christen HJ, Corry PC, Cowan FM, Cox H, D'Arrigo S, Dean J, De Laet C, De Praeter C, Dery C, Ferrie CD, Flintoff K, Frints SG, Garcia-Cazorla A, Gener B, Goizet C, Goutieres F, Green AJ, Guet A, Hamel BC, Hayward BE, Heiberg A, Hennekam RC, Husson M, Jackson AP, Jayatunga R, Jiang YH, Kant SG, Kao A, King MD, Kingston HM, Klepper J, van der Knaap MS, Kornberg AJ, Kotzot D, Kratzler W, Lacombe D, Lagae L, Landrieu PG, Lanzi G, Leitch A, Lim MJ, Livingston JH, Lourenco CM, Lyall EG, Lynch SA, Lyons MJ, Marom D, McClure JP, McWilliam R, Melancon SB, Mewasingh LD, Moutard ML, Nischal KK, Ostergaard JR, Prendiville J, Rasmussen M, Rogers RC, Roland D, Rosser EM, Rostasy K, Roubertie A, Sanchis A, Schiffmann R, Scholl-Burgi S, Seal S, Shalev SA, Corcoles CS, Sinha GP, Soler D, Spiegel R, Stephenson JB, Tacke U, Tan TY, Till M, Tolmie JL, Tomlin P, Vagnarelli F, Valente EM, Van Coster RN, Van der Aa N, Vanderver A, Vles JS, Voit T, Wassmer E, Weschke B, Whiteford ML, Willemsen MA, Zankl A, Zuberi SM, Orcesi S, Fazzi E, Lebon P, Crow YJ. Clinical and molecular phenotype of Aicardi-Goutieres syndrome. *Am J Hum Genet.* 2007 Oct;81(4):713-25. Epub 2007 Sep 4.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17846997>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2227922/>
- Rigby RE, Leitch A, Jackson AP. Nucleic acid-mediated inflammatory diseases. *Bioessays.* 2008 Sep;30(9):833-42. doi: 10.1002/bies.20808. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18693262>
- Shaban NM, Harvey S, Perrino FW, Hollis T. The structure of the mammalian RNase H2 complex provides insight into RNA-NA hybrid processing to prevent immune dysfunction. *J Biol Chem.* 2010 Feb 5;285(6):3617-24. doi: 10.1074/jbc.M109.059048. Epub 2009 Nov 18.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19923215>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2823502/>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/RNASEH2A>

Reviewed: July 2010

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services